ARUP's Laboratory Test Directory

KIT (D816V) Mutation by PCR, Fluid : 0040137

Mnemonic: KIT-F

Methodology:	Polymerase Chain Reaction
Performed:	Sun-Sat
Reported:	2-7 days
Specimen Required:	Collect: One 5 mL lavender (EDTA) or pink (K_2EDTA). Storage/Transport Temperature: 5 mL whole blood **OR** 3 mL bone marrow at 2-8°C. (Min: 1 mL) Remarks: Do not freeze. Unacceptable Conditions: Clotted specimens. Stability (collection to initiation of testing): Ambient: 24 hours; Refrigerated: 5 days; Frozen: Unacceptable

ARUP Consult®

Disease Topics

Mast Cell Disease

Patient DNA is isolated and subjected to allele-specific PCR amplification. The reaction uses an oligonucleotide primer set specific for the exon 17 of *KIT* on chromosome 4, and an allele-specific primer that specifically initiates amplification from the allele containing the point mutation in codon 816. Each assay includes a positive control reaction using DNA from a plasmid that contains the *KIT* D816V mutation and a negative control using placental DNA. The *KIT* gene sequences are present in the normal human genome and serve as a control for PCR in the assay.

Interpretive Data:	PCR products are analyzed by electrophoresis and UV transillumination of ethidium bromide stained gels. Results of this test must always be interpreted in the context of morphologic and other relevant data, and should not be used alone for diagnosis of malignancy. The D816V mutation can be detected if tumor represents at least 0.3% of cells. This test is performed pursuant to an agreement with Roche Molecular Systems, Inc. Refer to Statement B under Testing Information at http://www.aruplab.com.
CPT Code(s):	83891 Isolation; 83898 x2 Amplification; 83914 Mutation identification; 83894 Gel separation; 83912 Interpretation and report. Additional CPT code modifiers may be required for procedures performed to test for oncologic or inherited disorders.
Cross References:	Asp816Val, Fluid, CKIT, Fluid, D816V, Fluid, Mast Cell Disease, Fluid, Systemic Mastocytosis, Fluid

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